Holoprosencephaly with Severe Hydrocephalus

AUTHOR(S):

ISHIKAWA, MASATSUNE; HANDA, HAJIME; OSAKA, KUNIHIKO; MORI, KOREAKI; MATSUDA, ISAO

CITATION:

ISHIKAWA, MASATSUNE ...[et al]. Holoprosencephaly with Severe Hydrocephalus. 日本外科宝函 1980, 49(1): 119-128

ISSUE DATE:

1980-01-01

URL:

http://hdl.handle.net/2433/208402

Arch Jap Chir 49 (1), 119~128, Jan., 1980

症　　例

Holoprosencephaly with Severe Hydrocephalus

MASATSUNE ISHIKAWA, HAJIME HANDA, KUNIHIKO OSAKA,
KOREAKI MORI and ISAO MATSUDA*

Department of Neurosurgery, Kyoto University Medical School
*Department of Neurosurgery, Shiga University of Medical Science
Received for Publication, Dec., 10, 1979

Introduction

Holoprosencephaly is a congenital brain anomaly retaining various patterns of embryonic prosencephalon[2)10)]. Usually holoprosencephalic infants have micro — or trigonocephaly[2)]. However, macrocephaly due to hydrocephalus can be occurred in holoprosencephaly. Osaka et al[6)]. noted that hydrocephalus with holoprosencephaly comprised about 10% of all congenital hydrocepalus in Japan.

Median facial anomaly can predict the presence of holoprosencephaly as indicated by DeMyer and Zeman[2)]. However, this characteristic facial anomaly may not be present in semilobar and even in alobar types of holoprosencephaly[6)]. In such circumstances an appropriate neuroradiologic study is necessary to establish the diagnosis.

In this report, two holoprosencephalic babies with hydrocephalus, not associated with facial anomaly, were presented. Differential diagnosis from severe hydrocephalus and hydranencephaly is discussed.

Case report

Case 1.

A 1-month-old girl was admitted to Kyoto University Hospital because of enlarged head. The patient was delivered by cesarean section due to cephalopelvic disproportion. Facial anomaly was not noted. On admission, head circumference was 49cm and sun-set phenomenon was noted. CT scan (Fig. 1) showed some dilatation of the ventricles, which seemed to be composed of one large CSF space.

There was a space with CSF-density in the midoccipital region, which may be the

Key Words : Holoprosencephaly, Hydrocephalus, Facial anomaly, Macrocrania, CT scan, Angiography.
索引語：前脳症，水頭症，顔面奇形，巨頭症，CT スキャン，血管撮影.
Present Address : Department of Neurosurgery, Kyoto University Medical School, Sakyo-ku, Kyoto, 606, Japan.

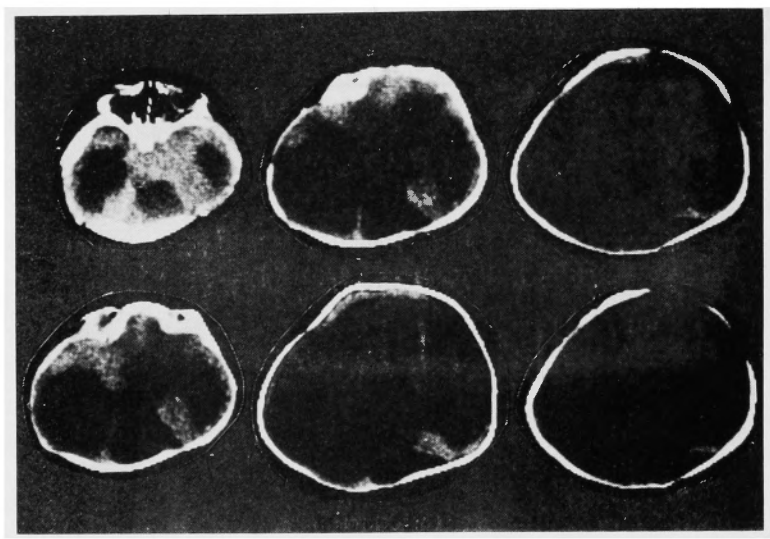


Fig. 1. Case 1. CT scan showing a large monoventricle. Note the CSF-density space in the midoccipital region. No falx can be seen.

dorsal sac in holoprosencephaly. Brain substance was seen only in the basal and frontal regions. On the higher slices the falx could not be seen. Left internal carotid angiogram (Fig. 2-A) showed the single anterior cerebral artery. In addition to the middle cerebral artery of normal origin, the midline portion of anterior cerebral artery gave rise not only to the ipsilateral middle cerebral artery, but also to the contralateral one. The left posterior cerebral artery was stretched and displaced beyond the midline. On the lateral view (Fig. 2-B), the anterior cerebral artery ran along the skull without undulation. The branches of bilateral middle cerebral arteries were stretched and did not form the sylvian triangle. Many perforating arteries could be seen. Right internal carotid angiogram showed the same findings except for lacking of the middle cerebral artery of normal origion. In the venous phase, a large avascular area involving almost whole hemisphere was observed on both sides. The superior sagittal sinus (Fig. 2-C) was present, but not the inferior sagittal sinus. On the left side (Fig. 2-D), draining veins from the thalamic region were noted. The vein from the basal part of brain ran backward along the pyramis ridge, which may be the ventral diencephalic vein in the embryonic stage. The vein from the dorsal aspect of thalamus ran posteriorly near the midline to the superior sagittal sinus. In the arterial phase of vertebral angiogram, the dense undivided thalamic image was seen and posterior circulation appeared normal other than the displacenent of lelt posterior cerebral arteries. In the venous phase (Fig. 3-A, B), bilateral veins from the dorsal aspect of thalamus ran posterosuperiorly to unite one large vein. Markedly dilated petrosal veins and superior petrosal sinuses were noted and the veins around the midbrain were well opacifeid.

Case 2.

A 5-day-old boy was admitted due to large head.

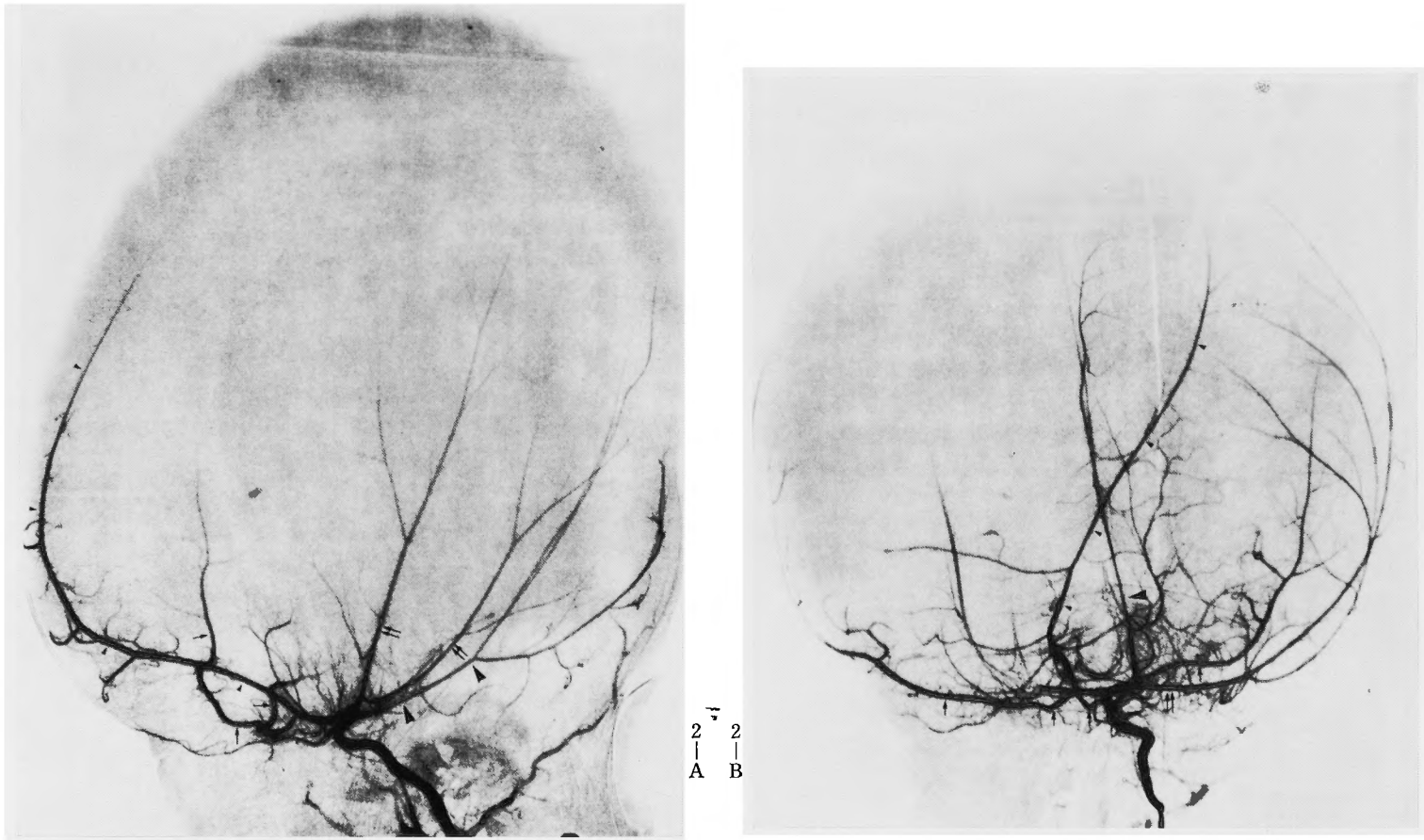
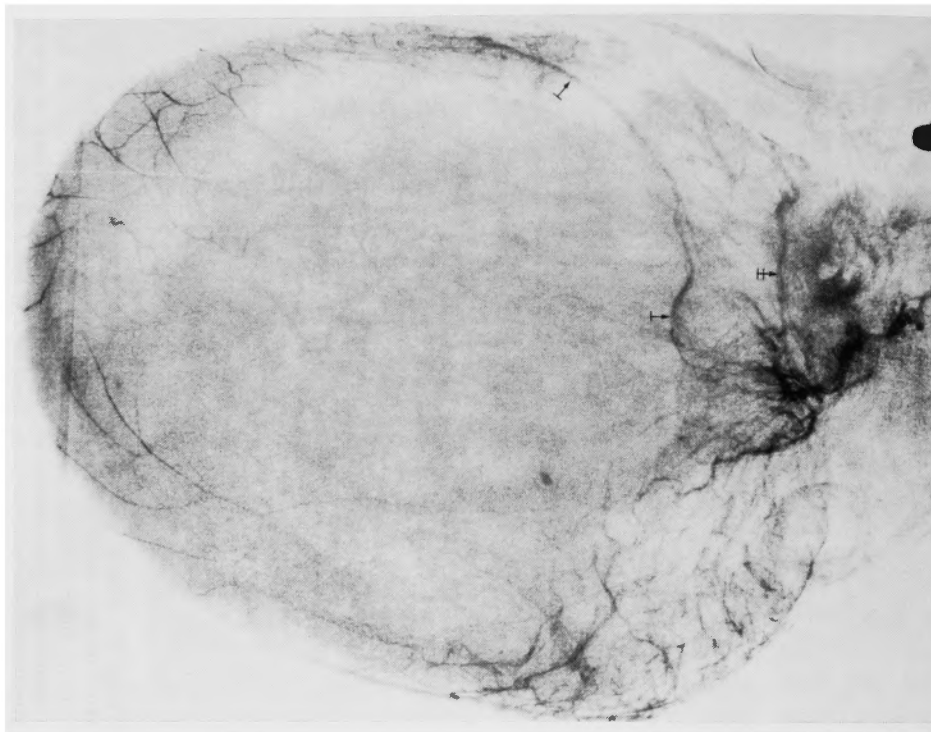
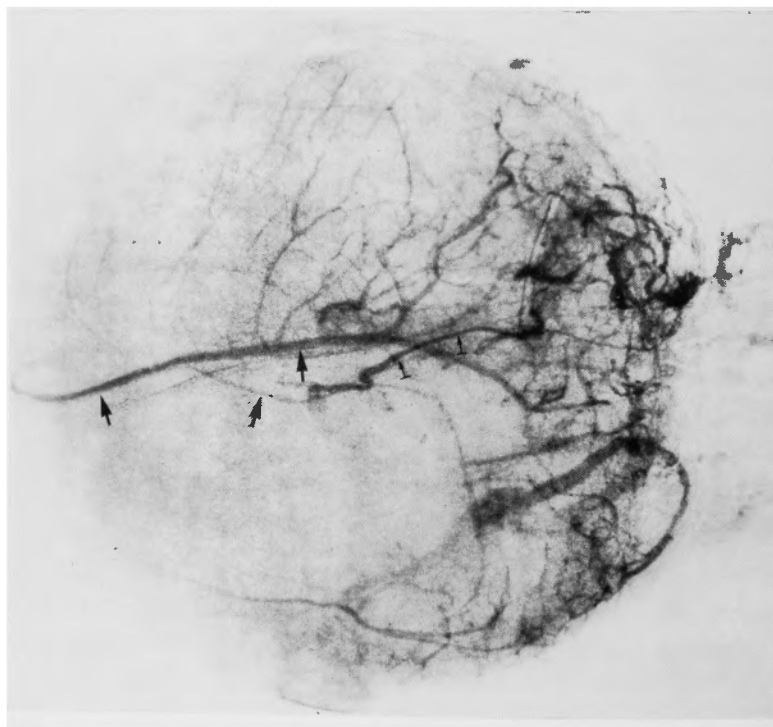


Fig. 2. Case 1. Left internal carotid angiogram. Arterial phase. A : A-P view. B : Lateral view. Azygos anterior cerebral artery (▲) ran close to the skull. From it, middle cerebral arteries (↑) were branched off on both sides. Another branch of left middle cerebral artery (↑↑) was derived from the bifurcation of left internal carotid artery. Posterior cerebral artery (△) was stretched. There were numerous fine branches in the diencephalon. Venous phase. C : A-P view. D : Lateral view. The superior sagittal sinus (↟) was composed of several large veins and posterior half of which became thin. There were numerous draining veins in the basal part of brain on both sides. There was a vein (↧) from the dorsal aspect of thalamus running backward near the midline. Another vein (↧) from the thalamus ran backward along the petrous fidge. A large avascular area wes seen.

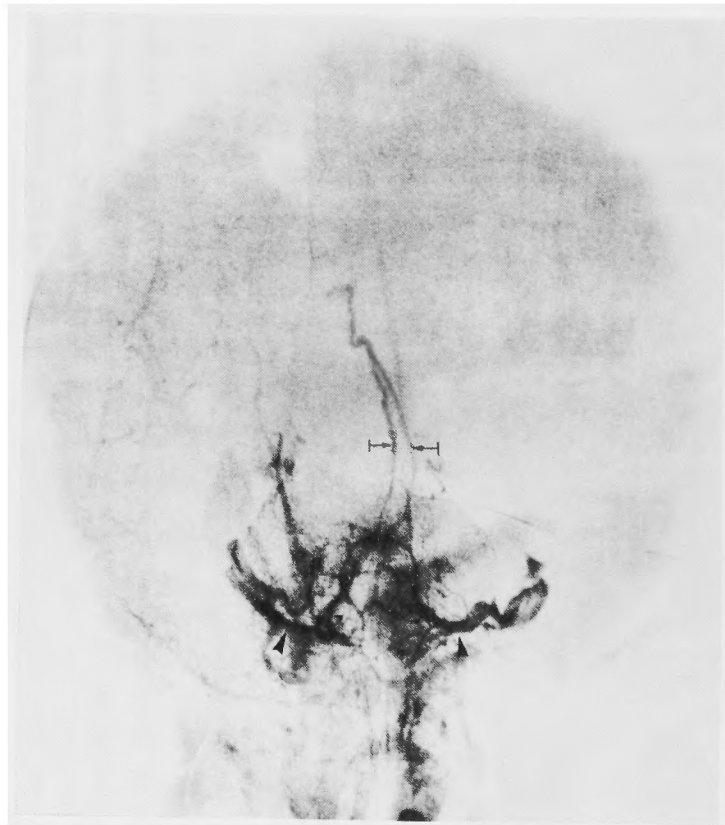


2—D



2—C

3—A



3—B

Fig. 3. Case 1. Left vertebral angiogram. A : Lateral view. B : Towne view. The draining veins (↓) from the dorsal aspect of thalamus ran posterosuperiorly to unite one large vein. Veins in the posterior fossa were well opacified. Especially, petrosal veins (▲) were well developed.

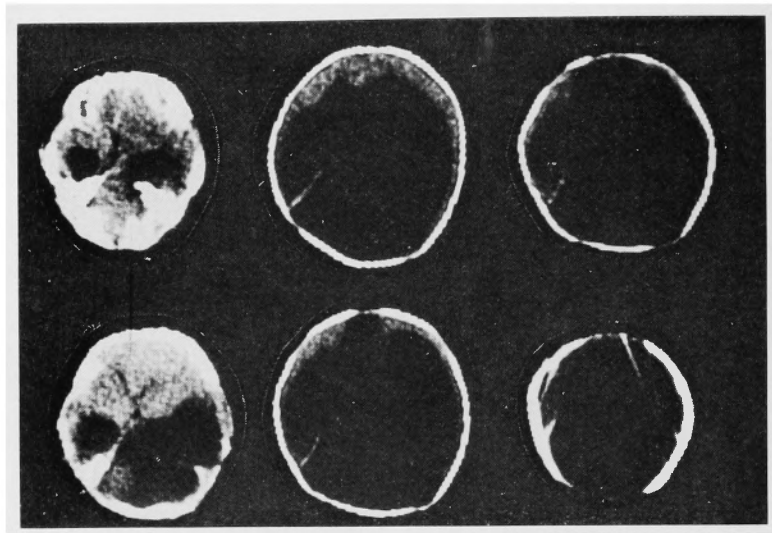


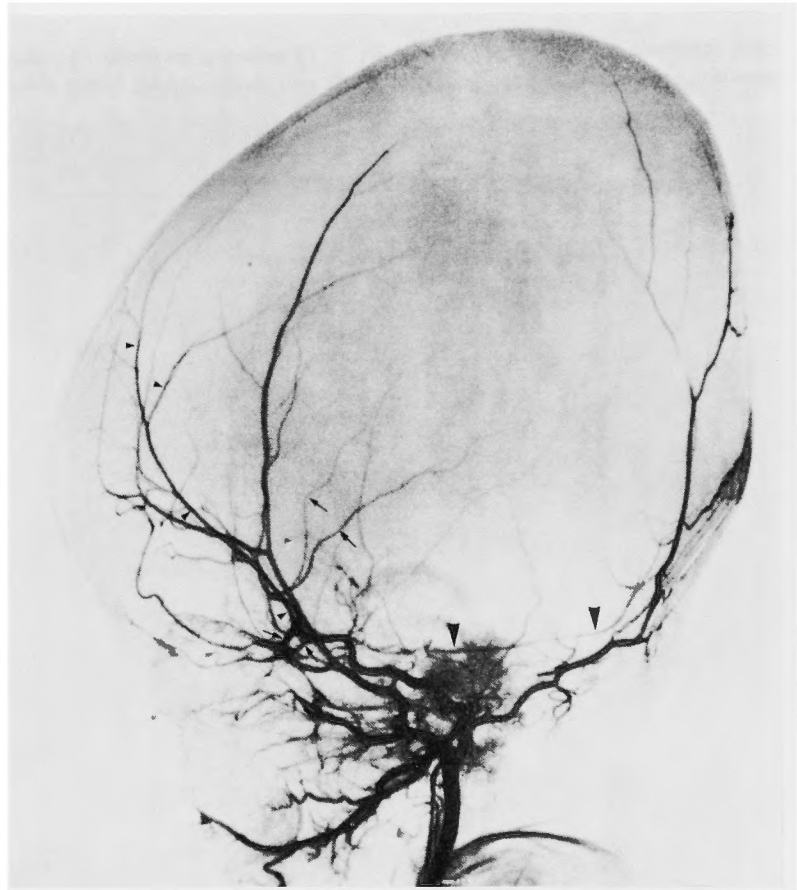
Fig. 4. Case 2. CT scan showing a large monoventricle with thin peripheral rim of cortex. No fals was seen, but there was a thin band extending from left pyramis up to parietal region.

He was delivered by cesarean section because of cephalopelvic disproportion. His head circumference on admission was 44.3cm. and sun-set phenomenon was present. No apparent facial anomaly was noted. CT scan (Fig. 4) revealed a markedly dilated CSF-density area suggesting monoventricle. Brain substance was present only in the basal and frontal regions. The falx was not identified on the midline. There was a thin band extending from the left pyrmis up to the parietal region. Right carotid angiogram (Fig. 5-A, B) showed the azygos anterior cerebral artery and their distal branches were stretched bilaterally away from the midline. The middle cerebral artery arose from the bifurcation of internal carotid artery, but no sylvian triangle could be outlined. Vertebral angiogram (Fig. 6-A) revealed the basilar artery took more horizontal course than usual, then it made an acute angle with the posterior cerebral artery which was displaced downward. In the venous phase (Fig. 6-B), there were three different venous drainages in the thalamic region ; anteroinferior, posteroinferior and posterosuperior drainages. The patient died 8 days after admission. Autopsy disclosed abscence of the corpus callosum and the medial cortex of brain, partial separation of the cerebral hemisphere, monoventricle, aqueductal stenosis and hypoplastic falx extending from the right petrous pyiramid to the occipital region. These findings indicate a semilobar holoprosencephaly with aqueductal stenosis.

Discussion

CT scan is of great help in evaluating various types of hydrocephalus. CT appearance of holoprosencephaly[1)9)] includes a single ventricle, fused thalamus, absence of the septum pellucidum, posterior extension of the midline CSF-density area and abscence or remnant

5—A



5—B

Fig. 5. Case 2. Right common carotid angiogram. A : A-P view. B : Lateral view. Azygos anterior cerebral artery (▲) was noted. Right middle cerebral artery (↑) branched from the bifurcation of internal carotid artery. Right posterior cerebral artery (▲) was markedly stretched and displaced downward.

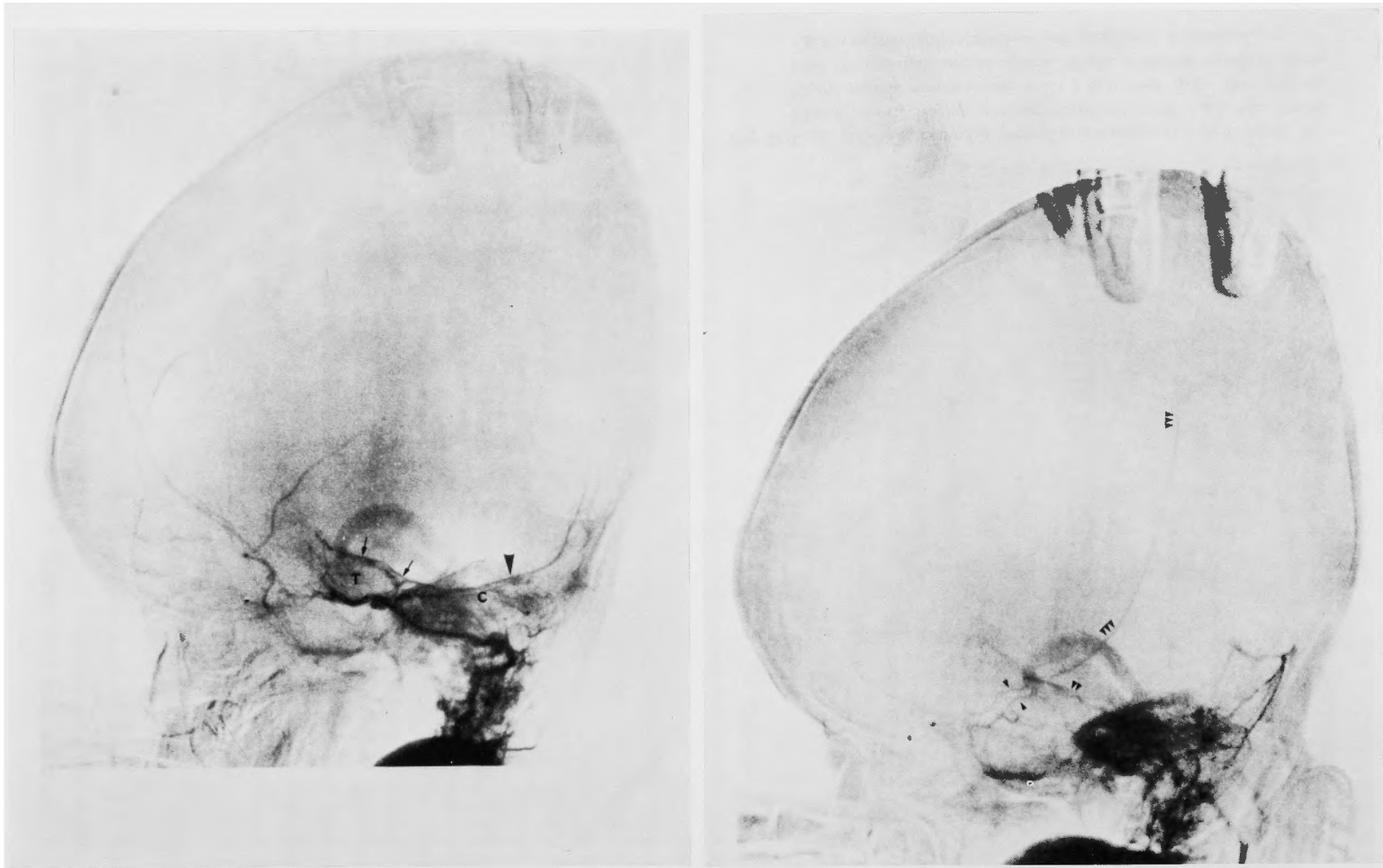


Fig. 6. Case 2. Vertebral angiogram. A : Late arterial phase. B : Venous phase. Basilar artery took more horizontal course than usual. Posterior cerebral artery (▲) was displaced downward and whose branches (↑) opacified thalamus (T). There were three different drainages from the thalamus (▲, ▲▲, ▲▲▲) C : Cerebellum.

of the falx and tentorium cerebelli. On CT scan, visualization of the falx with or without contrast enhancement is important[9], because it is abscent or hypoplastic in holoprosencephaly, but present in others. It is also useful for demonstrating facial anomaly[1], which is especially common in alobar holoprosencephaly. Although these findings on CT scan lead to the diagnosis of holoprosencephaly, alobar or semilobar holoprosencephaly with severe hydrocephalus are at times difficult to differentiate from severe hydrocephalus and hydranencephaly[1][9]. The differential diagnosis is mandantory from therapeutic standpoint because hydrocephalus can be treatable.

Angiography is useful for differentiating these disorders. In hydranencephaly, there are no or markedly diminished branches of the anterior and middle cerebral arteries, whearas, these branches are strectched, but still present in severe hydrocephalus. In holoprosencephaly, angiographic findings[3][4][5][8][11] are reportedly characterized by the azygos anterior cerebral artery, no pericallosal artery, no sylvian triangle formed by branches of the middle cerebral artery, abscence of the inferior sagittal sinus, abscence of the deep veins and remnant of the diencephalic veins in embryonic stage. Our cases also show these findings. In addition to the ventrolateral drainage from the diencephalon[5], in our Case 1, superoposteriorly running veins were noted. They originated from the dorsal aspect of thalamus bilaterally, running back in the almost midline, then probably into the superior sagittal sinus. They may be the same as "depressed internal cerebral vein" described by MAKI et al[4]. Since the roof of the third ventricle does not sink deep into the interhemispheric fissure in holoprosencephaly, the internal cerebral veins, which lie in the dorsal diencephalic roof, does not sink[5]. Thus these veins are unlikely to be the internal cerebral veins. The thalamus in the embryonic stage is drained bilaterally by tributaries of the dorsal and ventral diencephalic veins, which ran laterally to drain into the lateral sinus[7]. Thus, the dorsal diencephalic vein is also unlikely. Although it is difficult to define them in view of the vascular pattern of human embryo, there does exist the veins draining superoposteriorly from the thalamus and they seemed to run below the dorsal sac.

Since hydrocephalus can be treatable, while prognosis is poor in hydranencephaly and abobar/semilobar holoprosencephaly, precise diagnosis is urgent. CT scan is useful in evaluating these disorders. Angiography is also useful because holoprosencephaly retains various degrees of the vascular pattern of embryonic stage.

References

1) Byrd SE, Harwood-Nash DC, et al : Computed tomography evaluation of holoprosencephaly in infants and children. J Comput Assist Tomogr **1** : 456-463, 1977.
2) DeMyer W, Zeman W : Alobar holoprosencephaly (Arhinencephaly) with median cleft lip and palate : Clinical, electroencephalographic and nosologic consideration. Confin neurol **23** : 1-36 1963.
3) Harwood-Nash DC, Fitz CR : Neuroradiology in infants and Children Mosby, St. Louis 1976.
4) Maki Y, Kumagi K : Angiographic features of alobar holoprosencephaly. Neuroradiology **6** : 270-274, 1974.
5) Osaka K, Sato N, et al : Dysgenesis of the deep venous system as a diagnostic criterion for holoprosencephaly. Neuroradiology **13** : 231-238, 1977.
6) Osaka K, Matsumoto S : Holoprosencephaly in neurosurgical practice. J Neurosurg **48** : 787-803,

1978.
7) Padget D : The development of the cranial venous system in man from the viewpoint of comparative anatomy. Contr. Embryol. Carneg Instn **36** : 79-140, 1957.
8) Swett H, Nixon G : Agenesis of the corpus callosum with interhemispheric cyst. Radiology **114** : 641-645, 1975.
8) Weisberg L, Nice C : Cerebral Computed Tomography. A Text-Atals (Saunders, Philadelphia 1978).
9) Yakovlev P : Pathoarchitectonic studies of cerebral malformations. III. Arrhinencephalies (Holotelencephalies) J Neuropathol Exp **18** : 22-55, 1959.
10) Zingesser L, Schechter M : Angiographic and pneumoencephalographic features of holoprosencephaly. Am J Roentgenol **97** : 561-174, 1966.

和文抄録

著明な水頭症を伴った前脳症

京都大学医学部脳神経外科学教室
石川 正恒，半田 肇，寺阪 邦彦，森 惟明
滋賀大学医学部脳神経外科学教室
松田 功

著明な水頭症を伴った2例の前脳症を報告した．2例とも前脳症に特有な顔面奇形を伴っていなかった．神経放射線学的な観点から類似疾患との鑑別につき述べた．